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Goltz Syndrome with Mild Intellectual Disability and Excoriation (Skin-picking) - A Case Report

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Abstract **Background:** Goltz syndrome is a rare X-linked genetic syndrome of PORCN gene abnormality. It occurs predominantly in females while showing lethality for males. Most of the cases reported in the literature are bilateral with developmental defects in skin, skeletal, dentition, ocular and soft tissue. **Case Presentation:** Ms. X, 18-year old unmarried short statured female presented with symptoms of mild intellectual disability and excoriation, ocular colobomas, skin lesions, malocclusion of dentition, syndactyly, facial asymmetry and low set ears. **Conclusion:** This case is reported due to its rarity and highlights the importance of the psychiatric evaluation in detecting mild intellectual disability along with excoriation (skin-picking) in a characteristic Goltz syndrome.

Keywords: *goltz syndrome, Yale Brown Obsessive Compulsive Scale (YBOCS), Diagnostic and Statistical manual of mental disorders (DSM-5), International Classification of diseases: Clinical Descriptions and Diagnostic Guidelines (ICD-10)*

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1. Introduction

The first case report of focal dermal hypoplasia syndrome (FDHS) was reported by Jessner, 1921 [1]. Lieberman in 1935 described it as "atrophoderma linearis maculosa et papillomatosis congenitalis" [2]. It is a rare X-linked disorder of dominant gene with variable expressivity and abnormality of PORCN gene, a regulator of Wnt signalling [3]. Most of the cases are related to females (less than 300) and lethal for males as described by Goltz in 1962 [4]. However, Fernandez, 1975 reviewed the literature which included 55 cases, of which five i.e., 10% [5] and12% (15/125) of Hall's cases were males [6]. This syndrome is characterized by dysplasia of mesodermal and ectodermal structures with resultant defects in the skin, bone, eyes and soft tissues. Gorlin et al., 1990 reported mild mental retardation [7]. However, neurological abnormalities are invariably described in literature as meningomyelocoele, hydrocephalus, Arnold-Chiari malformation and absent corpus callosum [8]. Ophthalmic manifestations of FDHS occur in 40% of the cases as ocular-colobomas, strabismus, microphthalmus, hypertelorism, ectropion, ptosis, conjuctival papilloma, corneal clouding, aniridia, anophthamia etc., [9,10,11]. To the best of our knowledge, no similar case of Goltz syndrome with mild intellectual disability and excoriation (skin-picking) has been reported in the available literature.

2. The Case

Ms. X, 18-year-old unmarried female, born out of non-consanguineous marriage and having delayed developmental milestones presented to the Department of Psychiatry, Government Medical College and Rajindra Hospital, Patiala with persistent, repetitive attempts to stop the symptoms of excoriation (skin-picking) that resulted in skin lesions on the bilateral upper/ lower limbs for the last seven years. These symptoms were gradual in onset and worsened after her menarche, which was at the age of 11 years. She gets relief from these symptoms only when blood oozes out of her skin. These activities were extended on the skin of close relatives in spite of their resistance and the time spent was 3 to 4 hours daily. There was no past history of any substance or drug abuse (e.g. cocaine etc.) which could have precipitated her symptoms. The aggravation and remission of her symptoms had no relation with time, place, person and meal. She denied hearing any voices, tactile hallucinations as in psychotic disorder or delusion, stereotypies, body dysmorphic disorder and intention to harm oneself in non suicidal self-injury. She was second in birth order and her elder brother was normal. On general physical examination, she was found to be thin built, short stature with triangular facies and right facial asymmetry and hypoplasia. (Figure 1) She had low set ears with deformed pinna but normal hearing. (Figure 2).

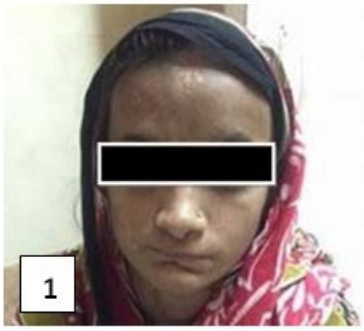

Figure 1. Right side facial asymmetry, hypoplasia with hypopigmented changes on forehead

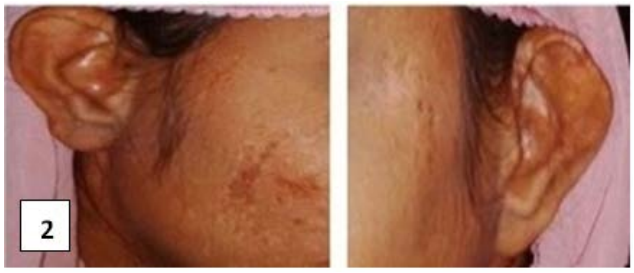

Figure 2. Low set ears with deformed pinna of both ears

Dental check-up showed maloccluded, hypoplastic irregular dentition. There was a notched right ala nasi. (Figure 3) On ophthalmological examination, best corrected visual acuity of both eyes was counting fingers close to face with convex lenses of +18 Dioptres and +20 Dioptre oculus dexter and oculus sinister respectively. Concomitant squint along with nystagmus was seen. Intraocular pressures of both eyes were 17 and 19 mm Hg.

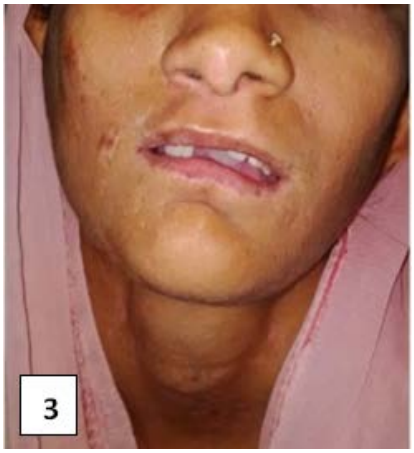

Figure 3. Irregular maloccluded dentition, notched right ala nasi with large nodule in front of neck

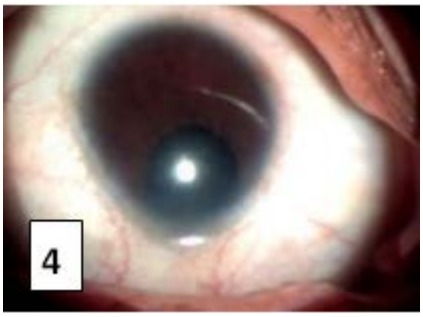

Figure 4. Microcornea with inferonasal iris coloboma oculus dexter

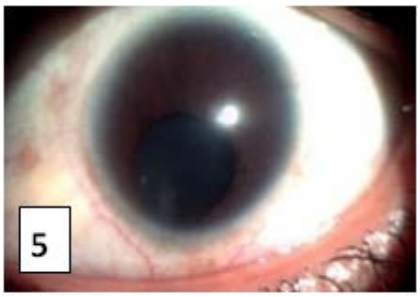

Figure 5. Microcornea, inferonasal iris coloboma with minimal cataract oculus sinister

Both eyes showed microphthalmos with microcornea of size of 9.5 mm vertically and 9 mm horizontally. Inferonasal iris colobomas were seen in both eyes along with minimal cataract in left eye. (Figure 4, Figure 5) Posterior segment evaluation revealed choroidal and disc colobomas with macular involvement in both eyes. Magnetic resonance imaging (MRI) scan, Electroencephalogram (EEG), ultrasonogarphy (USG) and echocardiography (ECG) were normal.

However, patient refused skin biopsy. A-Scan revealed axial length of 18 and 17.79 mm for right and left eye respectively. Cutaneous examination revealed multiple-linear atrophied hypo and hyperpigmented macules along the lines of Blaschko. She had patchy alopecia and sparse hair on eyebrows along with kyphosis and limb deformities in the form of polysyndactyly of both feet with nail dystrophy of left foot and syndactyly of index and middle fingers of right hand and nail dystrophy of middle finger of left hand. There were hypopigmentary changes on skin of both hands. (Figure 6, Figure 7) On mental status examination, her speech was normal, mood and affect showed anxiety.

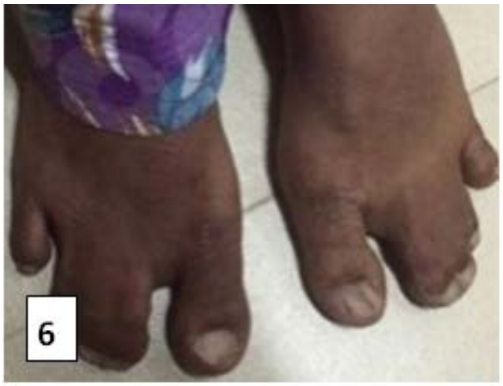

Figure 6. Polysyndactyly of both feet with nail dystrophy of left foot

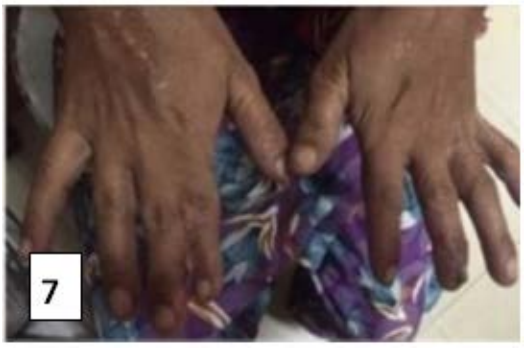

Figure 7. Syndactyly of right hand with nail dystrophy of middle finger of left hand

Thought process was persistent, repetitive and circumstantial. Thought content was of obsession-compulsive and related disorders e.g., "Excoriation (skin-picking) disorder." Her judgement was impaired; insight to illness was of grade-3 and Intelligence quotient (IQ) 63 with disability of 47%. Assessment of Disability in Persons of Mental Retardation scale (ADPMR) [12] showed score of 9 with moderate degree global disability in the range of 40-70%. The multiple axis classification of DSM-5/ICD-10 [13,14] showed diagnosis of Goltz syndrome (Q82.8) with mild Intellectual disability [317 (F70)] and Excoriation (skin- picking) disorder [698.4 (L98.1)]. Yale Brown Obsessive Compulsive Scale (YBOCS) [15] showed scores of 24 indicating severe illness and after 8 weeks score of 7. Clinical Global Impression [16] – Severity (CGI-S) scores were 6 and 2, respectively. She was treated with sertraline 50 mg per day and gradually titrated in the increment of 50 mg per week to the target level of 200 mg per day in divided doses. After 8 weeks, YBOCS and Clinical Global Impression – improvement (CGI-I) scores were 7 and 2 respectively; indicating much improvement after therapeutic intervention with minimal side effects and subsequent follow-up with other multiple disciplines for reconstructive surgeries/ management.

3. Discussion

Goltz syndrome (FDHS) is characterised by dysplasia of mesoectodermal structures having a locus at Xp11.23. [8]. Skin manifestations follow developmental lines known as Blaschko lines. It begins with inflammatory phase followed by desquamation, blistering and crusting. Hair may be brittle and sparse and nails poorly developed. [11,18] It should not be confused with Gorlin-Goltz syndrome which is entirely a different entity, a nevoid basal cell carcinoma [19]. In this case, skin of the patient showed lesions of hypo/hyper-pigmentation and telangiectasia that followed Blaschko lines.

There are many ocular abnormalities in Goltz syndrome e.g., anophthalmia, aniridia, strabismus, nystagmus, subluxation of lens, colobomas of the iris, choroids, retina and optic nerve etc., [10]. However, in the present case, there was exotropia, nystagmus, microphthalmos, microcornea, inferonasal iris, choroidal and disc colobomas with macular involvement of both eyes.

Dental abnormalities include rognathism, agenesis of the teeth, under development of the mandible, malocclusion, enamel defects with caries etc., [16]. In our case, there was maloccluded and hypoplastic irregular dentition.

Skeletal abnormalities e.g., short stature, kyphosis, scoliosis, spina-bifida occulta, pointed chin, asymmetry of the face, claw hands, syndactyly, clinodactly, polydactyl etc. are reported in literature [18]. Furthermore, this patient was of short stature, facial asymmetry, syndactyly, kyphosis and low set ears with deformed pinna.

A few case reports describe mental deficiency and seizures [8] without delineating whether it is congenital or acquired, their degree of severity and co-morbid psychiatric, neurological and/or medical illness. This case had mild intellectual disability with IQ of 63 and percentage disability of 47% and excoriation in Goltz syndrome. In the present case, the diagnosis of Goltz syndrome was made on the clinical grounds of facial asymmetry, dental, skeletal and ocular abnormalities as there was no facility for genetic screening.

4. Conclusion

The management of Goltz syndrome requires multidisciplinary approach. Psychological and intellectual issues should be addressed by the psychiatrists for their management and rehabilitation. This case is a rarity as it presents with mild intellectual disability and excoriation (skin-picking).

Acknowledgement

Nil.

Conflict of Interest

None Declared.

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